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A systematic review of issues around antenatal screening and prenatal diagnostic testing for genetic disorders: women of Asian origin in western countries

ABSTRACT

Antenatal screening has become standard practice in many countries. However, not all pregnant women choose to be tested. In the UK, the incidence of some birth defects is found to be higher in babies of Asian women than in those of women from other ethnic groups, while there is some evidence suggesting that ethnic minorities, especially Asian women, are less likely to undergo antenatal screening and prenatal diagnosis, the reasons for which are unclear. This paper aims to identify and describe the literature on issues around antenatal screening and prenatal diagnostic testing for genetic disorders among women of Asian descent in western countries. The Medline, CINAHL, ASSIA, and PsycInfo databases were searched for the period of 1995 and 2010. Twenty-one studies met the inclusion criteria and were therefore reviewed. In general, Asian women were found to hold favourable attitudes towards testing. However, they reported a poorer understanding of testing than white women and not being offered a test, and were less able to make informed choices. Asian women in the UK and Australia were found to be less likely than their white counterparts to have undergone prenatal diagnosis, while such differences were not found in the USA and Canada. The equity of access to quality antenatal care, alongside comprehensive well thought out antenatal screening programmes, can be assured if strategies are in place which actively involve all ethnic groups and take account of social and cultural appropriateness for the population served. An understanding of broad factors that inform women's decision-making on test uptake would help health professionals provide women and their families with more culturally sensitive information and support that they may additionally need in order to make more informed choices.

Keywords: Asian, genetic testing, minority ethnic clients, pregnancy, screening, systematic review

What is known about the topic

- Antenatal screening is standard practice in many countries.
- Not all pregnant women choose to undertake antenatal screening or prenatal diagnosis when they are offered it.
- Variations do exist in test uptake by ethnic group in some countries.

What this paper adds

- Asian women have difficulties in accessing antenatal screening and prenatal diagnostic testing, have a poor knowledge of testing, and are less able to make informed choices around test uptake.
- Asian women in the UK and Australia tend to be less likely than white women to undergo antenatal screening and prenatal diagnosis.
- Antenatal services need to take account of social and cultural appropriateness for the population served.

INTRODUCTION

Antenatal screening has become standard practice in many countries. In the UK, the National Screening Committee advises on all aspects of screening, makes nation wide policies and supports implementation of screening programmes (http://www.screening.nhs.uk/). The antenatal and newborn screening programme was launched in 2003 free of charge by the National Health Service, emphasising fairness, equality and test standards. The guidelines on this programme were supported by the Department of Health's Genetics White Paper (DH 2003), the National Institute for Health and Clinical Excellence guidance on antenatal care (NICE 2008), and the national service framework for children, young people and maternity services (DH 2004). A range of screening options are offered to pregnant women, including screening for infectious diseases and some genetic disorders caused by problems with either chromosomes (e.g. Down syndrome) or genes (e.g. sickle cell diseases and thalassaemia).

Down syndrome is a genetic condition caused by trisomy 21 and associated with intellectual disability, typical facial features and other medical problems (http://www.ncbi.nlm.nih.gov/omim). Antenatal screening for this condition is carried out through a blood test (maternal serum screening) and an ultrasound scan. Sickle cell diseases and thalassaemia are serious recessively inherited conditions that affect haemoglobin (http://ghr.nlm.nih.gov/). Antenatal screening for these conditions involves blood testing a woman for carrier status early in pregnancy and then her partner if she is a carrier. Screening tests for a condition provide an estimate of the risk of a pregnancy being affected and provide parents with information to enable them to make informed choices about definitive testing (Grant 2005, Tapon 2010). Using either amniocentesis or chorionic villus sampling, prenatal diagnostic testing can provide women and their families with reassurance that their baby has no detected abnormality, give parents time to prepare for the arrival of a baby with special needs, or allow parents to consider the termination of an affected baby. Prenatal diagnosis for a genetic condition can also be offered on a basis of advanced maternal age and a family or personal history of a genetic disorder (Tapon 2010).

Consanguineous marriage is common in some Asian populations in their homeland and overseas. It is estimated that at least 55% of British Pakistanis are married to their first cousins (Darr & Modell 1988, Rowlatt 2005). It is well known that consanguinity increases the likelihood of birth defects, including some recessive genetic disorders, such as sickle cell diseases and thalassaemia (Darr & Modell 1988, Stoltenberg *et al.* 1999, Obeidat *et al.* 2010). In the UK, the incidence of some birth defects is found to be higher in babies of Asian women than in those of women from other ethnic groups (Office for National Statistics 2004). Some birth defects with a genetic component may be identified by antenatal screening and prenatal diagnostic testing. There is some evidence suggesting that ethnic minorities are less likely than white women to undergo such tests in some countries (Park *et al.* 2007, Fransen *et al.* 2009a, b, 2010). Reviewing literature on UK social and ethnic inequalities in this area, Rowe *et al.* (2004) also found that minority ethnic women, especially those of South Asian origin, were less likely than white women to receive antenatal screening/diagnosis for Down syndrome and haemoglobin disorders and less likely to take up that offer. However, the reasons why such inequalities exist are unclear.

Countries like the UK, the USA and Canada have become multicultural societies with many diverse ethnic groups. The UK 2001 census indicated that about 4.6 million people from ethnic minorities resided in the country, making up 7.9% of the total population (Office for National Statistics 2005). Of these, 50.3% identified themselves as people of Asian origin, with Indians (22.7%) being the largest of all minority ethnic groups, followed

by Pakistanis (16.1%). This figure does not include Chinese people, a separate category on the census, who represented 5.3% of the non-white population. The US 2000 census showed that people of Asian descent represented 4.3% of the total US population, while in Canada, approximately 10% of the total population identified themselves as people of Asian origin in the 2006 census (US Census Bureau 2004, Statistics Canada 2010).

Little research on the topic of antenatal screening and prenatal diagnosis has been carried out with Asian populations in western countries. In the UK, the majority of findings in this area are based on research conducted with white populations (e.g. Williams *et al.* 2005, Crockett *et al.* 2008, Skirton & Barr 2010), while in the USA, racial and ethnic group based research in this area tends to compare white and low-income Hispanic or black populations (e.g. Browner *et al.* 1996, Gavin *et al.* 2004, Park *et al.* 2007, Li *et al.* 2008, Nakata *et al.* 2010). The multicultural nature of these countries may challenge traditional approaches to the delivery of antenatal services, as practices and patterns of health care developed for the general population may be, to some extent, culturally inappropriate for a population of mixed ethnic backgrounds (Atkin *et al.* 1998, Tsianakas & Liamputtong 2002a). UK government health policy, such as the Single Equality Scheme, stresses the need to ensure equality and diversity in health, social care and public health (DH 2009). The current antenatal screening policy also highlights the importance of providing all women with sufficient information to enable them to make informed choices (NICE 2008). The lack of available information about women of Asian origin around this matter makes it difficult for health professionals to facilitate informed decision-making among this group.

METHODS

Aim

This paper seeks to identify, describe and critique literature on issues around antenatal screening and prenatal diagnostic testing for genetic conditions among women of Asian origin residing in western countries.

Design

A literature review was undertaken, following the Centre for Reviews and Dissemination principles of conducting systematic reviews (CRD 2008).

Search methods

Literature in this area was retrieved from the electronic databases: Medline, CINAHL, ASSIA and PsycInfo, using the following keywords in various combinations:

- antenatal, prenatal, perinatal, maternal or pregnan*
- and test*, screen*, diagnos*, genetic*, chromosom*, congenital, sickle cell, thalass?emia, Down* syndrome, h?emoglobin* or cystic fibrosis
- and Asian*, race, ethnic*, Indian*, Pakistan*, Bangladeshi*, Chinese, Japanese, Korean* or Melanesia*

Inclusion criteria

The inclusion criteria were as follow:

- Sample: primarily women of Asian descent who lived in a western country
- Types of study: primary research and audits
- Language: English
- Publication dates: between 1995 and 2010 to review the most current literature

Exclusion criteria
Papers were excluded if they:

- were literature reviews or discussion articles.
- focused on health professionals or Asian community members rather than Asian women.
- did not provide a separate breakdown of results by ethnic group.

Search outcomes

In total, 1384 references were identified initially. A flow diagram of the selection process is shown in Figure 1, adapted from Moher *et al.* (2009). Only 21 studies satisfied the inclusion criteria and were therefore reviewed.
INSERT FIGURE 1 HERE

Quality appraisal

Given the heterogeneous nature of the studies included in terms of design, sample size, sampling strategies and methods of data collection, a quality score was not calculated. All studies that met the inclusion criteria were therefore included, but characteristics and quality of the studies were discussed.

Data extraction

The data extracted consisted of: bibliographic information, aim, sample, design, data collection, year(s) of screening, policy at the time, and key findings. These are summarised in Table 1 and Table 2. The data extraction was performed by single reviewer, so was the study assessment.

Synthesis

A narrative synthesis of the extracted data was carried out, as the studies included were too diverse to conduct a quantitative meta-analysis or qualitative meta-ethnography (CRD 2008).

FINDINGS

Characteristics and quality of the studies are reported first, followed by access to antenatal screening, access to prenatal diagnostic testing and issues around informed consent.

Characteristics and quality of the studies

The studies were conducted in the UK (13), USA (4), Australia (2) and Canada (2). The conditions being screened for or diagnosed mainly included Down syndrome (Table 1), or haemoglobin disorders (Table 2).

Seven studies applied quantitative approaches using self-reported questionnaires to collect data, ten were audits using data from medical records, three were qualitative studies, and one used a mixed method design. Only one study used a large random sample of respondents across regions, and most studies were limited by small numbers of respondents overall or in some comparison groups. The overall response rates appear to be high, ranging from 62% to 84%. It is difficult to know whether those who returned questionnaires were representative of their groups. It was reported in one study that non-responders were more likely to be born outside the UK (31% vs. 17%; Rowe *et al.* 2008a). In another study, women who were illiterate in written English were not invited to take part (Dormandy *et al.* 2005). The quality of clinical records seems to be poor. In some cases, healthcare providers

failed to document information on ethnicity, reproductive history, country of birth, or risk factors for foetal abnormality, such as consanguinity and family history of a genetic condition (Halliday *et al.* 1999, Hamilton & Maresh 1999, Learman *et al.* 2003, Dormandy *et al.* 2008). In one study, the women's ethnic origin was defined by religion (Hamilton & Maresh 1999). This is questionable, as it is unclear whether the differences reported for ethnic origin were due to ethnicity or religion. It is difficult to know how women's ethnicity had been classified in other studies.
INSERT TABLE 1 HERE

Access to antenatal screening

Down syndrome

The offer of antenatal screening for Down syndrome was reported in two UK studies (Hamilton & Maresh 1999, Rowe *et al*. 2008a). In a national survey on women's experiences of antenatal care, despite a universal offer policy, Asian women were significantly less likely than their white counterparts to report being offered the screening, even after adjusting for levels of deprivation, age and parity (84% vs. 89%, $OR_{adj} = 0.61$, $P = 0.02$; Rowe *et al.* 2008a). Similar results were reported in a review of retrospective case-notes of women who booked for antenatal care in 1991 at seven hospitals in England (Hamilton & Maresh 1999). Data from medical records would be more reliable than those from participant recall if the quality of recording is satisfactory. At two hospitals, the records on screening offered were generally poor, while at the remaining five hospitals, it was recorded that significantly fewer Asian women eligible for the screening (aged 37 years or older at delivery) were offered it compared to white women. The notes might not reflect accurately what had happened due to poor quality of recording; however, the findings did suggest some inequalities of access to screening.

Screening uptake was reported in five UK studies, with ethnic variations found. In two prospective hospital-based surveys of women attending antenatal care in England, the uptake was significantly lower in Asian women, who were either born in the UK or elsewhere, than that in white women (Chilaka *et al*. 2001, Dormandy *et al*. 2005). Similar results were reported in two audits and one national survey (Gilbert *et al.* 1996, Ford *et al.* 1998, Rowe *et al.* 2008a).
INSERT TABLE 2 HERE

Haemoglobin disorders

The uptake of antenatal screening for haemoglobin disorders was examined in one UK study (Dormandy *et al.* 2008). Of 1441 women from various ethnic groups attending antenatal care, 67% were screened for sickle cell diseases and thalassaemia, and the proportion screened did not vary by ethnic group.

In general, the findings suggest some inequalities of access to antenatal screening for Down syndrome in the UK, but not for haemoglobin disorders.

Access to prenatal diagnostic testing

Down syndrome

The uptake of prenatal diagnostic testing for Down syndrome was reported in seven studies. It was recorded in three UK audits that Asian women were less likely to be tested than white women although following screening, they were found to be at higher risk of

carrying a baby with the condition (Gilbert *et al.* 1996, Ford *et al.* 1998, Hamilton & Maresh 1999). A lower uptake rate of Asian women was also reported in an Australia audit where women aged 37 years or older at delivery were offered prenatal diagnosis for the condition (Halliday *et al.* 1995). However, studies carried out in the USA and Canada challenged these findings, where the diagnosis was offered on a basis of advanced maternal age, family history or subsequent to a high risk identified by antenatal screening (Kuppermann *et al.* 1996, 2006, Mueller *et al.* 2005, Saucier *et al.* 2005). Asian women were found to be as likely as white women to be tested and the uptake was significantly higher in both white and Asian women than that in black women and Latinas. The possible reasons for these inconsistencies may include variations in policy on testing across countries and in samples in terms of size and characteristics.

Haemoglobin disorders

The offer of antenatal screening for thalassaemia was reported in one UK audit (Modell *et al*. 2000). The analysis included 138 UK women who had a pregnancy affected by thalassaemia between 1990 and 1994. It was recorded that Asian women (Pakistani, Indian and Bangladeshi) were significantly less likely than other women to be offered antenatal screening for the condition during their first pregnancy or all pregnancies (35% vs. 73%, 53% vs. 86%, respectively). Also, in a study of 783 Chinese people living in Canada, 5% of respondents were identified as alpha-thalassaemia carriers and 1.7% as beta-thalassaemia carriers (Young *et al.* 1999). A significant proportion of couples were not identified as carriers prior to or early in a pregnancy. Of the 19 couples who underwent a diagnostic test for alpha-thalassaemia, five were offered the test late in pregnancy and of the 17 couples who undertook a diagnostic test for beta-thalassaemia, three had already had a child with the condition before being identified as carriers.

The uptake of prenatal diagnostic testing for haemoglobin disorders was investigated in two UK audits (Modell *et al*. 1997, 2000). Analysing data from the thalassaemia module of the Confidential Enquiry into Counselling for Genetic Disorders between 1990 and 1994, Modell *et al.* (2000) found that the uptake by UK Pakistani couples was 73% (35/48) in the first trimester and 39% (11/28) in the second trimester. In another audit, a breakdown of results on prenatal diagnostic testing for sickle cell diseases by ethnic group were unavailable, but variations in the uptake of prenatal diagnosis for thalassaemia were recorded, at 9% (15/168) in Bangladeshis, 28% (147/522) in Pakistanis, 53% (151/284) in Indians, and 94% (488/518) in Cypriots (Modell *et al*. 1997).

In summary, Asian women especially those in the UK tended to be less likely to have undergone prenatal diagnosis for Down syndrome or haemoglobin disorders.

Issues around informed consent

Informed consent was measured in only one study, where respondents who had a good knowledge of screening testing for Down syndrome and favourable attitudes towards screening actually underwent testing were classified as making informed choices about screening for the condition (Dormandy *et al.* 2005). Fewer Asian women than white women were found to have made informed choices (20% vs. 56%, $\chi^2 = 64$, $P < 0.001$). Women's knowledge, attitudes and awareness in terms of antenatal screening/diagnosis were also explored by others (Chilaka *et al*. 2001, Tsianakas & Liamputtong 2002a,b, Hewison *et al.* 2007, Ahmed *et al.* 2006, 2008).

Knowledge of antenatal screening and prenatal diagnosis

Dormandy *et al.* (2005) used a measurement tool consisting of eight questions to assess women's knowledge of screening for Down syndrome, with correct answers to more than four questions classified as indicating good knowledge. Asian women scored significantly lower than white women (3.6 vs. 5.5, $F = 45$, $P < 0.001$). Similar findings were reported by Chilaka *et al*. (2001), where respondents' knowledge of Down syndrome was classified as good, fair or poor, according to the levels of their understanding of the condition as a chromosomal abnormality associated with significant mental disability and structural abnormality. Significantly more white women were found to have a good knowledge of the condition than both UK-born and foreign-born Asian women (51% vs. 31%, $P = 0.003$; 51% vs. 8%, $P < 0.001$, respectively). Of Asian women, those born in the UK had a better knowledge than those born elsewhere, as did those who were able to speak English compared to those who were not. Foreign-born Asian women were more likely to report having received no screening information than both UK-born Asian women and white women (38%, 32% and 18% respectively).

These findings may reflect communication barriers, which were also found in a qualitative study of Australian women from an Islamic background, most of whom were of Asian descent (Tsianakas & Liamputtong 2002a). Respondents reported that they had limited opportunities to receive information on antenatal screening/diagnosis for Down syndrome, due to language barriers and a lack of cultural appreciation among health care providers. Unsurprisingly, findings from a study of UK Pakistani women showed that most participants were unaware of the risk of procedure-related miscarriage associated with prenatal diagnosis (Ahmed *et al*. 2006).

Attitudes to antenatal screening and prenatal diagnosis

Dormandy *et al.* (2005) found that both Asian and white respondents, in general, were in favour of screening for Down syndrome and there were no differences in attitudes seen between the two groups. However, among respondents in favour of the screening, Asian women were less likely than white women to have been screened (45% vs. 75%, $\chi^2 = 17.5$, $P < 0.001$). The reason for this variation was unclear and differences in uptake by ethnic group were not found in women not in favour of the screening.

Two UK studies were conducted to explore attitudes of white and Pakistani women towards prenatal diagnosis for 30 genetic conditions and termination of pregnancy for foetal abnormality (Hewison *et al.* 2007, Ahmed *et al.* 2008). Pakistani women scored significantly higher on favourable attitudes to testing than white women ($P < 0.001$; Hewison *et al.* 2007). Findings from a qualitative study of 19 women showed that there were more similarities than differences in women's attitudes between the two groups, but Pakistani women tended to highlight the role of religion more in their decisions on declining testing or termination than white women (Ahmed *et al.* 2008). In the USA, Learman *et al.* (2003) found that Asian women's attitudes towards prenatal diagnosis for Down syndrome were more likely to be influenced by their family members, but little by health professionals. The role of religion and family was also reported by others (Tsianakas & Liamputtong 2002b, Ahmed *et al.* 2006).

Awareness of being tested

It appeared that not all women reported being informed of testing. Chilaka *et al*. (2001) found that 48% (118/245) of a UK sample recalled having had a blood test for Down syndrome during pregnancy, although they had received a counselling session prior to the

test. This figure varied from 28% in foreign-born Asian women to 38% in UK-born Asian women, and 66% in white women. Also, in a recent UK survey, among all respondents who had undertaken the screening for Down syndrome, 4% (109/2891) did not remember being offered it (Rowe *et al.* 2008a).

Similarly, Ahmed *et al*. (2005) found that 88% of Pakistani respondents said that they had not been asked for their consent for thalassaemia carrier testing and most respondents reported that they found it difficult to ask health professionals questions due to their lack of pre-test information and language barriers. The findings may be subject to self-report bias and recall errors or reflect poor communication, but it could be the case that the provision of inadequate screening information might also be a contributing factore.

In some studies, Asian women thought that screening for Down syndrome and thalassaemia was the routine in antenatal care (Tsianakas & Liamputtong 2002b, Ahmed *et al*. 2005, 2006). The need for consent was not stressed by the majority of UK Pakistani participants (Ahmed *et al.* 2005), while the importance of being informed, but not being pressured, was highlighted by some Asian women in Australia (Tsianakas & Liamputtong 2002b).

In summary, although Asian women in general were in favour of antenatal screening/diagnosis, they showed little knowledge of screening and the condition being tested for, and were less able or had little opportunity to make informed choices.

DISCUSSION

Despite limited research in this area, findings from existing literature have indicated that in general, Asian women in western countries hold favourable attitudes towards antenatal screening and prenatal diagnosis for genetic disorders (Dormandy *et al*. 2005, Hewison *et al.* 2007, Ahmed *et al.* 2008). In the UK and Australia, however, Asian women were found to be less likely than white women to have undergone an antenatal screening or diagnostic test for Down syndrome or haemoglobin disorders (Gilbert *et al*. 1996, Ford *et al*. 1998, Hamilton & Maresh 1999, Model *et al*. 1997, 2000, Chilaka *et al*. 2001, Dormandy *et al*. 2005, Rowe *et al*. 2008a). These findings are consistent with previous reviews of the UK literature in this area, with ethnic minorities, especially South Asian women, less likely to report being offered or to take up antenatal screening/diagnosis (Rowe *et al*. 2004, Rowe & Garcia, 2005). Inequalities in this matter are not unique to the UK. For example, in the Netherlands, women from Turkish and Surinamese backgrounds were found to have a poorer knowledge about Down syndrome than Dutch women, be less likely to undertake screening for the condition, and be less able to make informed choices about screening (Fransen *et al*. 2009a, b, 2010).

The findings of this review have also suggested that differences in uptake of prenatal diagnostic testing for Down syndrome between Asian and white women were not evident in the USA and Canada (Kuppermann *et al.* 1996, 2006, Mueller *et al.* 2005, Saucier *et al.* 2005). These findings may be caused by variations in policy across countries and within a country. In the UK, the antenatal and newborn screening programme has only been introduced in the last few years, while there is no such programme in the USA, Canada and Australia. Response bias, recall difficulties, quality of recording, variations in characteristics of the samples and a failure to control confounding variables in some studies, such as socio-demographic factors, may have also contributed to these findings.

Low uptake of Asian women may be explained in part by access difficulties, as demonstrated by reports from women who did not recall being offered antenatal screening and from medical records suggesting that Asian women were less likely than white women

to be offered a screening test (Modell *et al*. 1997, 2000, Hamilton & Maresh 1999, Rowe *et al*. 2008a). It might be possible that the actual uptake among Asian women was much lower due to two additional factors. First, late attendance needs to be considered. Findings from UK national surveys showed that about 5% of respondents booked their first antenatal appointment after 12 weeks' gestation, and 2% after 20 weeks' gestation, with Asian women, especially those who were born outside the UK, being more likely to start antenatal care late (Rowe *et al*. 2008a, b). Also in the UK, women from Pakistan, Indian and other minority ethnic backgrounds were found to have fewer antenatal visits than white women (Petrou *et al.* 2001). Similar results were reported in the USA and Netherlands (Kuppermann *et al.* 1996, Gavin *et al.* 2004, Alderliesten *et al.* 2007, Park *et al.* 2007). For women who do not start antenatal care until after 12 or 20 weeks' gestation and for those with reduced antenatal visits, their opportunity to be tested for genetic disorders may be reduced or missed.

Poor communication may also contribute to access difficulties. As highlighted in the literature and the media, language is a widely recognised barrier for ethnic minorities to accessing health and social care services, including antenatal care (Bowes & Domokos 1996, Ahmed *et al*. 2005, Ansari *et al.* 2009, Triggle 2010, Wright 2010). Issues around interpretation have also been frequently documented, such as those related to availability of interpreters, difficulties encountered in translating medical terms and ethical aspects surrounding the use of interpreters in medical care (Atkin *et al.* 1998, Rozario 2005, Mastrocola & Nwabineli 2009). Language barriers could also have an impact on women's opportunities to receive information and their understanding of the process of testing and the genetic condition being tested for (Chilaka *et al*. 2001, Tsianakas & Liamputtong 2002a, Ahmed *et al*. 2005, Fransen *et al.* 2009b). People with limited English skills may also not receive much information about inherited diseases from their family members, as reported in studies of British Asian families (Atkin *et al.* 1998, Shaw & Hurst 2009). Partners fluent in English, usually husbands, often withheld genetic information to protect their spouse from blame, stigma or feelings of marital insecurity (Shaw & Hurst 2009). It is unlikely that women would be able to make informed choices without receiving sufficient information from service providers and more additional information from their families. Language barriers may also be an underlying cause of low participation levels of ethnic minorities in research (e.g. Rowe *et al*. 2008a), although it is possible that they might be excluded from research due to their lack of fluency in English (e.g. Dormandy *et al*. 2005). Providing written information in a woman's first language and the use of interpreters may increase minority women's opportunity to participate in research (e.g. Chilaka *et al*. 2001, Kuppermann *et al.* 2006, Rowe *et al*. 2008a). However, written information would be of no use to people who are illiterate. This was found to be the case in some studies with British Asian people (Lindesay *et al*. 1997, Stone *et al*. 2008).

Findings of this review have also suggested the impact of factors other than access difficulties on test uptake, such as knowledge of testing, attitudes to testing and the influence of religion, faith and family members. However, it is not clear whether any of these are perceived of being more important. Belonging to a minority ethnic group does not explain why inequalities of access exist when the issue of ethnicity arises. Findings from research with women in general have suggested that women's decisions on antenatal screening and prenatal diagnostic testing can be associated with a wide range of other factors, such as demographic characteristics and personal experiences (e.g. Green *et al.* 2004, Alderdice *et al.* 2008, Crockett *et al.* 2008, Rowe *et al.* 2008a, France *et al.* 2011). The impact of some factors, such as religious beliefs, on individuals' attitudes towards prenatal diagnosis, termination of a pregnancy and uptake of tests are often complex, with faith often used

within a broader context of family, cultural and social relationships (Rozario 2005, Ahmed *et al.* 2008, Atkin *et al.* 2008). Focusing exclusively on ethnic differences could lead to inaccurate generalisations on women's decisions on testing, which can be influenced by diverse factors other than ethnicity. Failure to control confounding social and cultural variables could result in a causal correlation between ethnicity and a lack of test uptake.

Review limitations

This is the first review on issues around antenatal screening and prenatal diagnosis in a under-researched group. Some limitations need to be acknowledged. The review did not include grey literature. Some pertinent literature that is not published may have been omitted. Also, the review only included papers published in the English language. This possibly omitted some relevant studies published in other languages due to the fact that all studies reviewed were conducted in English-speaking countries. The selected studies for inclusion may be subject to selective bias due to subjective judgement of the sole reviewer. In future work, such a review could be strengthened by establishing a team comprising of members with different expertise. However, based on this review, a number of recommendations can be made.

Recommendations for practice and research

First, antenatal interventions should be culturally appropriate to the needs of people from diverse cultural and ethnic backgrounds. Informing women of all the tests available at their first antenatal visit can be very challenging for care providers (Conaty *et al.* 2005). The amount of information that a woman can take in is questionable, and this is likely to be further exacerbated if English is not her first language. Additional attention should be paid to women who were born abroad, as they are often disadvantaged by language barriers, lack of social support, and are likely to be unfamiliar with the healthcare system in their host country.

Second, future research needs to explore women's experiences within the social and cultural context within which prenatal testing decision-making takes place. The perspectives of women, their families and the health professionals providing antenatal care services need to be taken on board. While existing research tends to rely on quantitative methods, quantitative data should be complemented by qualitative data to gain an additional understanding of research in this area.

Lastly, as with research in general, issues pertaining to design, sample size, sampling strategies and response rates need to be addressed. Strategies, which may need to be agreed with research funding bodies, should be in place to expand the participation of ethnic minorities, especially those who are unable to speak English or who are illiterate. Also, more research in this area is needed to provide up-to-date evidence, since only two UK studies reviewed were conducted after the launch of the national antenatal and newborn screening programme.

CONCLUSION

This review provides some evidence suggesting ethnic inequalities in access to antenatal screening and diagnosis for genetic disorders among Asian women residing in western countries. In any multicultural country, health professionals need to provide antenatal care relevant to women and their families from all ethnic groups. The equity of access to quality

antenatal services, alongside comprehensive well thought out screening programmes, can be assured if strategies are in place which actively involve all ethnic groups and take account of social and cultural appropriateness for the population served. An understanding of broad factors that inform women's decision-making on test uptake would help health professionals provide women and their families with more culturally sensitive information and support that they may additionally need in order to make informed choices.

REFERENCES

Ahmed S., Green J. & Hewison J. (2005) Antenatal thalassaemia carrier testing: women's perceptions of 'information' and 'consent'. *Journal of Medical Screening* 12 (2), 69-77.

Ahmed S., Green J.M. & Hewison J. (2006) Attitudes towards prenatal diagnosis and termination of pregnancy for thalassaemia in pregnant Pakistani women in the North of England. *Prenatal Diagnosis* 26 (3), 248-257.

Ahmed S., Hewison J., Green J.M., Cuckle H.S., Hirst J. & Thornton J.G. (2008) Decisions about testing and termination of pregnancy for different fetal conditions: A qualitative study of European white and Pakistani mothers of affected children. *Journal of Genetic Counseling* 17, 560-572.

Alderdice F., McNeill J., Rowe R., Martin D. & Dornan J. (2008) Inequalities in the reported offer and uptake of antenatal screening. *Public Health* 122, 42-52.

Alderliesten M.E., Vrijkotte T.G.M., van der Wal M.F. & Bonsel G.J. (2007) Late start of antenatal care among ethnic minorities in a large cohort of pregnant women. *British Journal of Gynaecology* 114, 1232-1239.

Ansari W.E.l., Newbigging K., Roth C. & Malik F. (2009) The role of advocacy and interpretation services in the delivery of quality healthcare to diverse minority communities in London, United Kingdom. *Health & Social Care in the Community* 17 (6), 636-646.

Atkin K., Ahmed S., Hewison J. & Green J. (2008) Decision-making and ante-natal screening for sickle cell and thalassaemia disorders. *Current Sociology* 56 (1), 77-98.

Atkin K., Ahmad W.I. & Anionwu E.N. (1998) Screening and counselling for sickle cell disorders and thalassaemia: the experience of parents and health professionals. *Social Science & Medicine* 47 (11), 1639-1651.

Bowes A.M. & Domokos T.M. (1996) Pakistani women and maternity care: raising muted voices. *Sociology of Health & Illness* 18 (1), 45-65.

Browner C.H., Preloran M. & Press N.A. (1996) The effects of ethnicity, education and an informational video on pregnant women's knowledge and decisions about a prenatal diagnostic screening test. *Patient Education and Counseling* 27, 135-146.

Centre for Reviews and Dissemination (2008) *Systematic Reviews: CRD's Guidance for Undertaking Reviews in Health Care.* York Publishing Services Ltd, York.

Chilaka V.N., Konje J.C., Stewart C.R., Narayan H. & Taylor D.J. (2001) Knowledge of Down syndrome in pregnant women from different ethnic groups. *Prenatal Diagnosis* 21 (3), 159-164.

Conaty S.J., Cassell J.A., Harrisson U., Whyte P., Sherr L. & Fox Z. (2005) Women who decline antenatal screening for HIV infection in the era of universal testing: results of an audit of uptake in three London hospitals. *Journal of Public Health* 27 (1), 114-117.

Crockett R., Wilkinson T.M. & Marteau T.M. (2008) Social patterning of screening uptake and the impact of facilitating informed choices: Psychological and ethical analysis. *Health Care Analysis* 16, 17-30.

Darr A. & Modell B. (1988) The frequency of consanguineous marriage among British Pakistanis. *Journal of Medical Genetics* 25, 186-190.

Department of Health (2003) *Our Inheritance, Our Future: Realising the Potential of Genetics in the NHS* [WWW document]. URL http://www.dh.gov.uk/prod_consum_dh/groups/dh_digitalassets/@dh/@en/documents/digitalasset/dh_4019239.pdf, last accessed 27 April 2011.

Department of Health (2004) *National Service Framework for Children, Young People and Maternity Services: Maternity Services* [WWW document]. URL http://www.dh.gov.uk/en/Publicationsandstatistics/Publications/PublicationsPolicyAndGuidance/DH_4089101, last accessed 27 April 2011.

Department of Health (2009) *Single Equality Scheme 2009-2012* [WWW document]. URL http://www.dh.gov.uk/prod_consum_dh/groups/dh_digitalassets/documents/digitalasset/dh_100963.pdf, last accessed 27 April 2011.

Dormandy E., Michie S., Hooper R. & Marteau T.M. (2005) Low uptake of prenatal screening for Down syndrome in minority ethnic groups and socially deprived groups: a reflection of women's attitudes or a failure to facilitate informed choices? *International Journal of Epidemiology* 34 (2), 346-352.

Dormandy E., Gulliford M.C., Reid E.P., Brown K., Marteau T.M. & SHIFT Research Team. (2008) Delay between pregnancy confirmation and sickle cell and thalassaemia screening: a population-based cohort study. *British Journal General Practice* 58 (548), 154-159.

Ford C., Moore A.J., Jordan P.A. Bartlett W.A., Wyldes M.P., Jones A.F. & MacKenzie W.E. (1998) The value of screening for Down's syndrome in a socioeconomically deprived area with a high ethnic population. *British Journal of Obstetrics and Gynaecology* 105 (8), 855-859.

France E.F., Wyke S., Ziebland S., Entwistle V A. & Hunt K. (2011) How personal experiences feature in women's accounts of use of information for decisions about antenatal diagnostic testing for foetal abnormality. *Social Science & Medicine* 72 (5), 755-762.

Fransen M.P., Wildschut H.I., Vogel I., Mackenbach J.P., Steegers E.A. & Essink-Bot M.L. (2009a) Ethnic differences in considerations whether or not to participate in prenatal screening for Down syndrome. *Prenatal Diagnosis* 29 (13), 1262-1269.

Fransen M.P., Wildschut H.I., Vogel I., Mackenbach J.P., Steegers E.A. & Essink-Bot M.L. (2009b) Information about prenatal screening for Down syndrome: Ethnic differences in knowledge. *Patient Education and Counseling* 77, 279-288.

Fransen M.P., Essink-Bot M.L., Vogel I., Mackenbach J.P., Steegers E.A. & Wildschut H.I. (2010) Ethnic differences in informed decision-making about prenatal screening for Down's syndrome. *Journal of Epidemiology and Community Health* 64, 262-268.

Gavin N.I., Adams E.K., Hartmann K.E., Benedict M.B. & Chireau M. (2004) Racial and ethnic disparities in the use of pregnancy-related health care among Medicaid pregnant women. *Maternal and Child Health Journal* 8 (3), 113-126.

Gilbert L., Nicholl J., Alex S. *et al.* (1996) Ethnic differences in the outcome of serum screening for Down's syndrome. *British Medical Journal* 312, 94-95.

Grant S.S. (2005) Options for Down syndrome screening: what will women choose? *Journal of Midwifery & Women's Health* 50, 211-218.

Green J.M., Hewison J., Bekker H.L., Bryant L.D. & Cuckle H.S. (2004) Psychosocial aspects of genetic screening of pregnant women and newborns: a systematic review. *Health Technology Assessment* 8 (33), 1-119. [WWW document]. URL http://www.hta.ac.uk/fullmono/mon833.pdf, last accessed 27 April 2011.

Halliday J., Lumley J. & Watson L. (1995) Comparison of women who do and do not have amniocentesis or chorionic villus sampling. *Lancet* 345 (18), 704-709.

Hamilton S.M. & Maresh M.J. (1999) Antenatal screening by history taking - a missed opportunity. *Journal of Obstetrics and Gynaecology* 19 (1), 10-14.

Hewison J., Green J.M., Ahmed S., Cuckle H.S., Hirst J., Hucknall C. & Thornton J.G. (2007) Attitudes to prenatal testing and termination of pregnancy for fetal abnormality: a comparison of white and Pakistani women in the UK. *Prenatal Diagnosis* 27, 419-430.

Kuppermann M., Gates E. & Washington A.E. (1996) Racial-ethnic differences in prenatal diagnostic test use and outcomes: preferences, socioeconomics, or patient knowledge? *Obstetrics & Gynecology* 87 (5), 675-682.

Kuppermann M., Learman L.A., Gates E., Gregorich S.E., Nease R.F. Jr, Lewis J. & Washington A.E. (2006) Beyond race or ethnicity and socioeconomic status: predictors of prenatal testing for Down syndrome. *Obstetrics & Gynecology* 107 (5), 1087-1097.

Learman L.A., Kuppermann M., Gates E., Nease R.F. Jr, Gildengorin V. & Washington A.E. (2003) Social and familial context of prenatal genetic testing decisions: are there racial/ethnic differences? *American Journal of Medical Genetics. Part C, Seminars in Medical Genetics* 119C (1), 19-26.

Li D.K., Karlberg K., Wi S. & Norem C. (2008) Factors influencing women's acceptance of prenatal screening tests. *Prenatal Diagnosis* 28 (12), 1136-1143.

Lindesay J., Jagger C., Hibbett M.J., Peet S.M. & Moledina F. (1997) Knowledge, uptake and availability of health and social services among Asian Gujarati and white elderly persons. *Ethnicity & Health* 2 (1-2), 59-69.

Mastrocola E.L. & Nwabineli N.J. (2009) Antenatal services for ethnic minority women in South Tyneside. *British Journal of Midwifery* 17 (7), 418-423.

Modell B., Petrou M., Layton M. *et al.* (1997) Audit of prenatal diagnosis for haemoglobin disorders in the United Kingdom: the first 20 years. *British Medical Journal* 315 (7111), 779-784.

Modell B., Harris R., Lane B. *et al.* (2000) Informed choice in genetic screening for thalassaemia during pregnancy: audit from a national confidential inquiry. *British Medical Journal* 320 (7231), 337-341.

Moher D., Liberati A., Tetzlaff J. & Altman D.G. (2009) Preferred reporting items for systematic reviews and meta-analyses: the PRISMA statement. *British Medical Journal* 339, 332-339.

Mueller V.M., Huang T., Summers A.M. & Winsor S.H.M. (2005) The influence of risk estimates obtained from maternal serum screening on amniocentesis rates. *Prenatal Diagnosis* 25, 1253-1257.

Nakata N., Wang Y. & Bhatt S. (2010) Trends in prenatal screening and diagnostic testing among women referred for advanced maternal age. *Prenatal Diagnosis* 30, 198-206.

National Institute for Health and Clinical Excellence (2008) *Antenatal Care: Routine Care for the Healthy Pregnant Woman.* 2nd edn. [WWW document]. URL http://www.nice.org.uk/nicemedia/live/11947/40115/40115.pdf, last accessed 27 April 2011.

Obeidat B.R., Khader Y.S., Amarin Z.O., Kassawneh M. & Al Omari M. (2010) Consanguinity and adverse pregnancy outcomes: the north of Jordan experience. *Maternal and Child Health Journal* 14 (2), 283-289.

Office for National Statistics (2004) *The health of children and young people: chapter 11: Congenital anomalies.* [WWW document]. URL http://www.statistics.gov.uk/children/downloads/congen_anomalies.pdf, last accessed 20 January 2007.

Office for National Statistics (2005) *Ethnicity and Identity: Population Size.* [WWW document]. URL http://www.statistics.gov.uk/cci/nugget.asp?id=455

Park J.H., Vincent D. & Hastings-Tolsma M. (2007) Disparity in prenatal care among women of colour in the USA. *Midwifery* 23 (1), 28-37.

Petrou S., Kupek E., Vause S. & Maresh M. (2001) Clinical, provider and sociodemographic determinants of the number of antenatal visits in England and Wales. *Social Science & Medicine* 52, 1123-1134.

Rowe R., Garcia J. & Davidson L.L. (2004) Social and ethnic inequalities in the offer and uptake of prenatal screening and diagnosis in the UK: a systematic review. *Public Health* 118, 177-189.

Rowe R. & Garcia J. (2005) Ethnic minorities' access to antenatal screening. *British Journal of Midwifery* 13 (2), 101-104.

Rowe R., Puddicombe D., Hockley C. & Redshaw M. (2008a) Offer and uptake of prenatal screening for Down syndrome in women from different social and ethnic backgrounds. *Prenatal Diagnosis* 28, 1245-1250.

Rowe R., Magee H., Quigley M.A., Heron P., Askham J. & Brocklehurst P. (2008b) Social and ethnic differences in attendance for antenatal care in England. *Public Health* 122 (12), 1363-1372.

Rowlatt J. (2005) *The risks of cousin marriage* [WWW document]. URL http://news.bbc.co.uk/1/hi/programmes/newsnight/4442010.stm, last accessed 28 April 2011.

Rozario S. (2005) Genetics, religion and identity among British Bangladeshis: some initial findings. *Diversity in Health and Social Care* 2, 187-196.

Saucier J.B., Johnston D., Wicklund C.A., Robbins-Furman P., Hecht J.T. & Monga M. (2005) Racial-ethnic differences in genetic amniocentesis uptake. *Journal of Genetic Counseling* 4 (3), 189-195.

Shaw A. & Hurst J.A. (2009) 'I don't see any point in telling them': attitudes to sharing genetic information in family and carrier testing of relatives among British Pakistani adults referred to a genetic clinic. *Ethnicity & Health* 14 (2), 205-224.

Skirton H. & Barr O. (2010) Antenatal screening and informed choice: a cross-sectional survey of parents and professionals. *Midwifery* 26 (6), 596-602.

Statistics Canada (2010) *Population by Visible Minority Groups: 2006 Census* [WWW document]. URL http://www12.statcan.ca/census-recensement/2006/dp-pd/hlt/97-562/pages/page.cfm?Lang=E&Geo=PR&Code=01&Table=1&Data=Count&StartRec=1&Sort=2&Display=Page, last accessed 27 April 2011.

Stone M.A., Patel N., Daly H. *et al.* (2008) Using qualitative research methods to inform the development of a modified version of a patient education module for non-English speakers with type 2 diabetes: experience from an action research project in two South Asian populations in the UK. *Diversity in Health and Social Care* 5, 199-206.

Tapon D. (2010) Prenatal testing for Down syndrome: comparison of screening practices in the UK and USA. *Journal of Genetic Counseling* 19, 112-130.

Triggle N. (2010) *Vulnerable Pregnant Women 'Miss Out on Antenatal Care'* [WWW document]. URL http://www.bbc.co.uk/news/health-11381123, last accessed 27 April 2011.

Tsianakas V. & Liamputtong P. (2002a) What women from an Islamic background in Australia say about care in pregnancy and prenatal testing. *Midwifery* 18, 25-34.

Tsianakas V. & Liamputtong P. (2002b) Prenatal testing: the perceptions and experiences of Muslim women in Australia. *Journal of Reproductive and Infant Psychology* 20 (1), 7-24.

US Census Bureau (2004) *We the People: Asians in the United States* [WWW document]. URL http://www.census.gov/prod/2004pubs/censr-17.pdf, last accessed 27 April 2011.

Williams C., Sandall J., Lewando-Hundt G., Heyman B., Spencer K. & Grellier R. (2005) Women as moral pioneers? Experiences of first trimester antenatal screening. *Social Science & Medicine* 61, 1983-1992.

Wright C. (2010) *NHS 'Failing Those Who Don't Speak English'* [WWW document]. URL http://www.bbc.co.uk/news/health-10951417, last accessed 27 April 2011.

Young K.N., Wadsworth L.D., Langlois S. & Wilson R.D. (1999) Thalassemia carrier screening and prenatal diagnosis among the British Columbia (Canada) population of Chinese descent. *Clinical Genetics* 55, 20-25.

Table 1 Studies relating to Down syndrome (n=12)

Reference & country	Aim	Sample & response rate	Design & data collection	Year(s) of screening & policy at the time	Relevant key findings
Main focus: antenatal screening					
Chilaka *et al.* (2001) UK	To assess the level of awareness and understanding of Down syndrome in women from different ethnic groups	Women attending antenatal care at 1 Leicester hospital over a 3-month period N = 245 • White (117) • Non UK-born Asian (86) • UK-born Asian (32) • Other (10) Response rate: 82%	Prospective survey Self-administered questionnaire	Information on the year of data collection and policy at the time was not provided.	Asian women reported a poorer knowledge of Down syndrome than white women and lower uptake of the screening. Uptake was significantly higher in those with a good knowledge than those with a poor knowledge (58% vs. 23%, $P < 0.02$). Women's knowledge of the condition was associated with their ability to speak English.
Dormandy *et al.* (2005) UK	To assess whether the lower uptake of antenatal screening for Down syndrome by women from minority ethnic groups and socioeconomically disadvantaged groups reflected their attitudes	Women receiving antenatal care at 2 hospitals N = 1499 • White (1286) • South Asian (104) • Black (62) • Other (36) • Ethnicity not recorded (11) Response rate: 84%	Prospective survey Self-administered questionnaire Laboratory records	2000 Testing for Down syndrome (double test) was offered to all women at 15-16 weeks of gestation at both hospitals.	Most women were in favour of screening and their attitudes did not differ by ethnic group ($F = 1.0$, $P = 0.37$). Asian women had a lower uptake rate (29% vs. 52%, $\chi^2 = 26$, $P < 0.001$) and a lower knowledge of the testing ($F = 45$, $P < 0.001$) than white women. Uptake-attitude consistency was

					lower in Asian women than that in white women (62% vs. 78%, $\chi^2 = 17.5$, $P < 0.001$) and Asian women were less likely to make informed choices (20% vs. 56%, $P < 0.001$).
Ford *et al.* (1998) UK	To assess the uptake of screening for Down syndrome in a socioeconomically deprived area with a high proportion of Asian women	Women attending antenatal care at 1 hospital and 1 clinic in Birmingham during a 4-year period N = 16 827	Retrospective audit Data from medical records	10/1992-12/1996 All women with singleton pregnancies who booked antenatal care before 21 weeks' gestation were offered screening for Down syndrome; those aged 35 or older at delivery had the option of a diagnostic test.	The estimated overall uptake of screening was 71%, which was lower in Asian women than in white women. Following screening, more Asian women were found to be at high risk of carrying a baby with Down syndrome than white women (8% vs. 5%), but it was recorded that they were less likely to have undergone prenatal diagnosis (35% vs. 67%).
Gilbert *et al*. (1996) UK	To explore ethnic differences in outcomes of screening for Down syndrome	Women attending antenatal care before 18 weeks' gestation in Oldham, England N = 9217 • White and other women (7562) • Women of Indian origin (1655)	Retrospective audit Data from medical records	2/1991-10/1993 All women booked antenatal care before 18 weeks of gestation were offered screening for Down syndrome.	Fewer Asian women were screened than white and other women (72% vs. 90%). Following screening, more Asian women were found to be at high risk of carrying a baby with Down syndrome (12% vs. 4%), but it was recorded that fewer had

					undergone diagnostic testing when offered (27% vs. 60%, $\chi^2 = 43.9$, $P < 0.01$).
Hamilton & Maresh (1999) UK	To examine the effectiveness of identifying and managing women at increased risk of foetal abnormality	Women attending antenatal care at 7 hospitals in 2 health regions over 1 year N = 19 895	Retrospective audit Data from medical records	1-12/1991 Screening or diagnostic testing for Down syndrome was offered at all the hospitals according to maternal age.	At 5 hospitals, it was recorded that Asian women were significantly less likely than white women to be offered screening for Down syndrome or undergo the screening when offered ($P < 0.05$).
Rowe *et al*. (2008a) UK	To compare offer and uptake of antenatal screening for Down syndrome in women from different social and ethnic backgrounds	Women 3 months after giving birth Random sample using birth registration data in England N = 2960 • White women • Asian women • Black women • Other Response rate: 62%	Survey Postal questionnaire	3/2006 Screening for Down syndrome was offered to all pregnant women.	Asian women were found to be significantly less likely than white women to report being offered the screening (84% vs. 89%, $OR_{adj} = 0.61$, 95% CI = 0.39-0.94, $P = 0.02$) or to undertake it when offered (59% vs. 69%, $OR_{adj} = 0.48$, 95% CI = 0.33-0.72, $P < 0.001$).
Main focus: prenatal diagnostic testing					
Halliday *et al.* (1995) Australia	To compare factors that affected uptake of prenatal diagnostic testing for Down syndrome	All women aged 37 years or older who gave births between 1988 and 1990 in the 'foetal diagnosis data collection' at Victoria Clinical Genetics Service N = 7111	Case review Data from clinical records	1988-1990 Amniocentesis and chorionic villus sampling were available to women aged 37 years or older at delivery.	It was recorded that women born in Asia and other non-English speaking countries were significantly less likely to have tested than those born in English speaking countries (OR = 0.47, 95% CI = 0.40-

					0.55; OR = 0.55, 95% CI = 0.47-0.64; respectively, *P* < 0.001).
Kuppermann *et al.* (1996) USA	To examine whether the use of prenatal diagnostic testing for Down syndrome varied by ethnic group	Women aged 35 years and older who attended antenatal care at the University of California by 20 weeks' gestation N = 238 • Latinas (33) • African-American women (24) • White women (120) • Asian women (61)	Case review Data from medial records	11/1993-12/1994 Prenatal diagnosis for Down syndrome was offered to all women aged 35 years or older at delivery	Asian women did not differ from white women in their uptake of prenatal diagnostic testing for the condition. It was recorded that white and Asian women were more likely to have tested than Latinas and African-American women (72%; 75%, OR_{adj}=1.77; 33%, OR_{adj}=0.28; 33%, OR_{adj}=0.33; respectively).
Kuppermann *et al.* (2006) USA	To identify predictors of prenatal genetic testing decisions and explore whether racial or ethnic and socioeconomic differences are explained by knowledge, attitudes and preferences	Pregnant women attending antenatal care by 20 weeks of gestation at 23 San Francisco Bay-area clinics and practices N = 827 • African-American women • Asian women • Latinas • White women Response rate: 64%	Prospective survey Questionnaire	1997-1998 Women younger than 35 years were generally offered antennal screening only as the initial test. Women aged 35 years or older were offered a choice between prenatal diagnostic testing and antenatal screening.	Among women aged 35 years and younger, there were no differences in the uptake of screening and diagnosis by ethnic group (OR = 1.22, *P* = 0.17). Among women aged 35 years and older, black women had lower odds of undergoing screening/diagnosis than Asian women, white women and Latinas (OR=0.21, 0.40, 0.41, respectively). Of all women, these with higher perceived risk of procedure-related miscarriage

					were less likely to undergo diagnostic testing ($P = 0.037$), those with higher value of testing information were more likely to undertake diagnosis ($P < 0.001$), and those with greater health care system distrust or higher levels of faith/fatalism were less likely to undergo any screening/diagnostic testing ($P = 0.045$, 0.035, respectively).
Learman *et al.* (2003) USA	To explore attitudes to prenatal diagnostic genetic testing and the role of external influences among pregnant women	Women attending antenatal care by 20 weeks of gestation at 23 San Francisco Bay-area clinics and practices N = 999 • African-American women (18%) • Asian women (26%) • Latinas (22%) • White women (34%) Response rate: 64%	Survey Questionnaire	1997-1998 Information on policy at the time was not provided.	There was little endorsement by any group of the statement that faith/religion influenced their prenatal diagnostic testing decisions. Few women reported being influenced by their family's feelings about having a child with Down syndrome, but Asian women were more likely to report such an influence than other women and were less likely to report the influence by health care professionals. Attitudes to diagnostic testing varied within each ethnic group.
Mueller *et al.*	To explore the	Women screened in a	Case review	10/1993-9/1998	Of 7.1% of women who

(2005) Canada	influence of risk estimates obtained from antenatal screening for Down syndrome on uptake of diagnostic testing	Ontario maternal serum screening programme with a high risk screen result N = 16 792 • White women (11 980) • Asian women (2401) • Black women (1135) • Other (148) • Ethnicity not recorded (1128)	Data from laboratory and medial records	Maternal serum screening for Down syndrome was offered to all pregnant women.	received a high risk screen result, 65.7% had amniocentesis. It was recorded that Asian women were as likely as white women to undergo amniocentesis (67% vs. 66.6%, $P = 0.69$), but black women were less likely to be tested than both white and Asian women (48.6%, $P < 0.01$). Except for Asian and black women aged under 35, women were less likely to have been tested as their estimate risks decreased.
Saucier *et al.* (2005) USA	To investigate ethnic differences in uptake of diagnostic testing for Down syndrome	Women referred for amniocentesis at a hospital N = 157 • White women (33%) • African-American women (23%) • Hispanic women (22%) • Asian women (19%) • Other (4%) Response rate: 62%	Survey Questionnaire	8/2001-3/2002 Amniocentesis was offered to women according to advanced maternal age, abnormal serum screening, family history of a genetic disorder, or abnormal foetal ultrasound.	White, Asian and black women were more likely to undergo amniocentesis than Hispanic women (84%, 83%, 83% and 52%, respectively, $P = 0.003$).

Table 2 Studies relating to haemoglobin and others disorders (n=9)

Reference, country	Aim	Sample & response rate	Design & data collection	Year(s) of screening & policy at the time	Relevant key findings
Main focus: antenatal screening					
Ahmed *et al.* (2005) UK	To explore women's attitudes to informed consent for antenatal thalassaemia carrier testing and their perceived pre-test information needs	Pregnant women tested for thalassaemia carrier status in two cities in the North of England N = 146 • Pakistani (139) • White (5) • Black (2)	Mixed design Questionnaire and semi-structured interviews	9/1999-3/2001 Testing for thalassaemia was routinely offered to all pregnant women.	77.3% of women were unaware that they were tested for carrier status and 83.8% would have wanted to be informed. Most women received little or no pre-test information due to language barriers. They reported that they did not know enough about the test or condition to ask health professionals any questions and tended to think the testing was routine. 88.4% reported they were not asked for their consent for testing. Women stressed the importance of receiving information about the test, but not giving consent.
Dormandy *et al.* (2008) UK	To explore whether pregnant women had access to timely screening for sickle	Women attending antenatal care in 25 general practices from two primary care trusts	Cohort study Data from clinical and laboratory records	2005-2006 Screening for sickle cell and thalassaemia was offered to all	965 (67%) women undertook the screening before 26 weeks' gestation and the proportion screened did not

	cell diseases and thalassaemia	N = 1441 • North European • South and other European • Black African & African Caribbean • South and south-east Asian • Other • Mixed		pregnant women.	vary by ethnic group. Women's ethnic group was not associated with delay between pregnancy confirmation and the screening.
Main focus: prenatal diagnostic testing					
Ahmed *et al.* (2006) UK	Explore the attitudes of pregnant women towards prenatal diagnosis and termination of pregnancy for thalassaemia	Pregnant Pakistani Muslim women tested for thalassaemia carrier status N = 43	Qualitative design Semi-structured interviews	9/1999-3/2001 Testing for thalassaemia was routinely offered to all pregnant women	Most women were unaware of the risk of procedure-related miscarriage or the need to make decisions about diagnostic testing and termination of pregnancy following carrier testing. Most of them would have opted for prenatal diagnosis, emphasising the need to know whether their child had the condition. Women's families influenced their decisions on testing and termination or sometimes made the decision for them.
Ahmed *et al.* (2008)	To explore reasons for or against	Women with a child who had a genetic condition or	Qualitative design Semi-structured	Information on the year of data	There were more similarities than differences between the

UK	prenatal diagnostic testing and termination for foetal abnormality among white and Pakistani mothers of affected children	had terminated a pregnancy for a child with a genetic condition N = 19 • Pakistani women (10) • White women (9)	interviews & questionnaire	collection and policy at the time was not provided.	two groups. All women would have opted for termination for at least one of the 30 conditions listed. Pakistani women were more likely to highlight the role of religion in their decision-making than white women.
Hewison *et al.* (2007) UK	To compare the attitudes to prenatal diagnostic testing and termination of pregnancy for foetal abnormality	Women 6 weeks after delivery N = 420 • Pakistani women (198) • White women (222) Response rate: 65%	Survey Questionnaire	Information on the year of data was not provided. Screening for Down syndrome, sickle cell diseases and thalassaemia was offered to all pregnant women.	For most conditions, more than half of respondents wanted testing. Pakistani respondents held more favourable attitudes to prenatal diagnostic testing than white women ($P < 0.01$), but were less in favour of termination for foetal abnormality.
Modell *et al.* (1997) UK	To audit services for prenatal diagnosis for haemoglobin disorders	All UK cases of prenatal diagnosis for haemoglobin disorders between 1974 and 1994 N = 2068 • Cypriots • Pakistanis • Indians • Bangladeshis • Other	Retrospective audit Laboratory and clinical records	1974-1994 Screening was offered to all pregnant women who were not of northern European origin.	Uptake of diagnostic testing for thalassaemia was 9% (15/168) in Bangladeshis, 28% (147/522) in Pakistanis, 53% (151/284) in Indians and 94% (488/518) in Cypriots.
Modell *et al.* (2000)	To investigate informed choice in	All UK women identified as having a pregnancy	Retrospective audit	1990-1994 Screening was	The proportion of first and all pregnancies where prenatal

UK	antenatal screening and diagnostic testing for thalassaemia	affected by thalassaemia between 1990 and 1994 N = 138 • Cypriots (42) • Pakistanis (52) • Indians (26) • Bangladeshis (8) • Other (10)	Laboratory and clinical records	offered to all pregnant women who were not of northern European origin.	diagnosis was offered varied between British Cypriot and Pakistani couples (81% vs. 29%, $P < 0.0001$; 92% vs. 51%, respectively). Diagnostic testing in the first trimester was offered to 87% of Cypriot, but only to 50% of Pakistani couples with eligible pregnancies. Uptake by Pakistani couples was 73% (35/48) in the first trimester and 39% (11/28) in the second trimester.
Tsianakas & Liamputtong (2002a,b) Australia	To examine women's perceptions, experience and satisfaction in relation to antenatal care, antenatal screening and prenatal diagnostic testing	Islamic women living in Melbourne N = 15 • Lebanon (4) • Jordan (2) • Turkey (2) • Egypt (1) • Kuwait (2) • Malaysia (1) • Singapore (1) • Morocco (1) • Pakistan (1)	Qualitative method In-depth interviews	1999 Ultrasound and maternal serum screening were offered to all women. Those aged over 35 years were offered a diagnostic test, including chorionic villus sampling and amniocentesis.	In general, women had positive experiences with their care and were in favour of antenatal screening, particularly ultrasound, but some were ambivalent about amniocentesis. They tended to believed screening was the routine in their care and stressed the need to accept advice from their doctors as a 'normal mother'. Their decisions on screening/diagnosis were also influenced by their partners. Some reported having

					received insufficient information on antenatal screening and prenatal diagnostic testing due to language barriers and a lack of cultural appreciation among care providers.
Young et al. (1999) Canada	To identify whether routine screening for thalassaemia was indicated for the Chinese population in British Columbia, Canada	Patients of Chinese origin residing in British Columbia N = 783	Observational study Data from medial records	1/1991-6/1997 Antenatal screening for thalassaemis was not routinely offered.	Respondents who were identified as alpha-or beta-thalassaemia carriers were 5% and 1.7% respectively. 19 couples underwent prenatal diagnosis for alpha-thalassaemia in 25 pregnancies. Of these, 5 were referred late in pregnancy. 17 couples undertook diagnostic testing for beta-thalassaemia in 22 pregnancies. Three couples had already had a child with the condition before being identified as carriers.